# Figure 2

| Mutation | Exemplary Pool | 3' blocking group | Oligo type (Arm) | Sequence (5'-3') | $\varepsilon_{260}$ $M^{-1}$ $cm^{-1}$ |
|---|---|---|---|---|---|
| 2789+5G>A | 1 | none | invader | TTTGGTTGTGCTGTGGCTCCTTGGAAAGTGAT (SEQ ID NO:1) | 330800 |
| 2789+5G>A | 1 | hex | probe/DM | CGCGCCGAGGATATTCCATGTCCTATTGTG (SEQ ID NO:2) | 306500 |
| 2789+5G>A | 1 | none | synthetic target | CAATCTACACAATAGGACATGGAATATTCACTTTCCAAGGAGCCACAGCACAACCAAA (SEQ ID NO:3) | 667000 |
| R1162X | 1 | none | invader | GTTTACCTTCTGTTGGCATGTCAATGAACTTAAAGACTCT (SEQ ID NO:4) | 428000 |
| R1162X | 1 | hex | probe/DM | CGCGCCGAGGAGCTCACAGATCGC (SEQ ID NO:5) | 253000 |
| R1162X | 1 | none | synthetic target | TCAGATGCGATCTGTGAGCTGAGTCTTTAAGTTCATTGACATGCCAACAGAAGGTAAAC (SEQ ID NO:6) | 659000 |
| R347P | 4 | none | invader | CAGGGAAATTGCCGAGTGACCGCCATGT (SEQ ID NO:7) | 306600 |
| R347P | 4 | hex | probe/ER24 | ACGGACGCGGAGGGCAGAACAATGCAG (SEQ ID NO:8) | 318200 |
| R347P | 4 | none | synthetic target | CTCATTCTGCATTGTTCTGCCCATGGCGGTCACTCGGCAATTTCCCTGGG (SEQ ID NO:9) | 488000 |
| 1898+1G>A | 1 | none | invader | GACTCTCCTTTTGGATACCTAGATGTTTTAACAGAAAAAGAAATATTTGAAAGT (SEQ ID NO:10) | 619900 |
| 1898+1G>A | 1 | hex | probe/DM | CGCGCCGAGGATATGTTCTTTGAATACCTTACTTAT (SEQ ID NO:11) | 386500 |
| 1898+1G>A | 1 | none | synthetic target | ATAAGTAAGGTATTCAAAGAACATATCTTTCAAATATTTCTTTTTCTGTTAAAACATCTAGGTATCCAAAAGGAGAGTC (SEQ ID NO:12) | 904800 |
| 2184delA | 4 | none | invader | CCCCAAACTCTCCAGTCTGTTTAAAAGATTATTTTTTC (SEQ ID NO:13) | 393000 |
| 2184delA | 4 | hex | probe/DM | CGCGCCGAGGGTTTCTGTCCAGGAGACA (SEQ ID NO:14) | 305200 |
| del1507 | 1 | none | invader | GCTTTGATGACGCTTCTGTATCTATATTCATCATAGGAAACACCAAT (SEQ ID NO:15) | 509300 |
| del1507 | 1 | hex | probe/DM | CGCGCCGAGGAGATATTTTCTTTAATGGTGCC (SEQ ID NO:16) | 345200 |
| del1507 | 1 | none | synthetic target | GCCTGGCACCATTAAAGAAAATATCTTTGGTGTTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCC (SEQ ID NO:17) | 866600 |
| G85E | 4 | none | invader | GCCCTTCGGCGATGTTTTTTCTGGAGATTTATGTTCTATGT (SEQ ID NO:18) | 409100 |
| G85E | 4 | hex | probe/ER24 | ACGGACGCGGAGAAATCTTTTTATATTTAGGGGTAAG (SEQ ID NO:19) | 431700 |
| G85E | 4 | none | synthetic target | AGATCCTTACCCCTAAATATAAAAAGATTTCATAGAACATAAATCTCCAGAAAAAACATCGCCGAAGGGCATTA (SEQ ID NO:20) | 869300 |
| R117H | 3 | none | invader | AATCATAGCTTCCTATGACCCGGATAACAAGGAGGAACT (SEQ ID NO:21) | 443800 |
| R117H | 3 | hex | probe/DM | CGCGCCGAGGACTCTATCGCGATTTATCT (SEQ ID NO:22) | 304200 |
| R117H | 3 | none | synthetic target | ATGCCTAGATAAATCGCGATAGAGTGTTCCTCCTTGTTATCCGGGTCATAGGAAGCTATGATT (SEQ ID NO:23) | 681700 |
| R560T | 1 | none | invader | CATGAATGACATTTACAGCAAATGCTTGCTAGACCAATAATTAGTTATTCACT (SEQ ID NO:24) | 595000 |
| R560T | 1 | hex | probe/ER24 | ACGGACGCGGAGGTTGCTAAAGAAATTCTTGCT (SEQ ID NO:25) | 378100 |
| R560T | 1 | none | synthetic target | CAACGAGCAAGAATTTCTTTAGCAACGTGAATAACTAATTATTGGTCTAGCAAGCATTTGCTGTAAATGTCATTCATGTAAAA (SEQ ID NO:26) | 945400 |
| 3120+1G>A | 2 | none | invader | GCAATTTTGGATGACCTTCTGCCTCTTACCATATTTGACTTCATCCAGT (SEQ ID NO:27) | 496000 |
| 3120+1G>A | 2 | hex | probe/DM | CGCGCCGAGGATATGTAAAAATAAGTACCGTTAA (SEQ ID NO:28) | 397500 |
| 3120+1G>A | 2 | none | synthetic target | AGACATACTTAACGGTACTTATTTTTACATATCTGGATGAAGTCAAATATGGTAAGAGGCAGAAGGTCATCCAAAATTGCTATATC (SEQ ID NO:29) | 984000 |
| 3659delC | 2 | none | invader | GAGAGTTGGCCATTCTTGTATGGTTTGGTTGACTTT (SEQ ID NO:30) | 372300 |
| 3659delC | 2 | hex | probe/DM | CGCGCCGAGGGTAGGTTTACCTTCTGTTGG (SEQ ID NO:31) | 302800 |
| 3659delC | 2 | none | synthetic target | CATGCCAACAGAAGGTAAACCTACAAGTCAACCAAACCATACAAGAATGGCCAACTCTC (SEQ ID NO:32) | 679800 |
| A455E | 1 | none | invader | CCTGAAAGATATTAATTTCAAGATAGAAAGAGGACAGTTGTTGGT (SEQ ID NO:33) | 531000 |
| A455E | 1 | hex | probe/ER24 | ACGGACGCGGAGAGGTTGCTGGATCCA (SEQ ID NO:34) | 298100 |
| A455E | 1 | none | synthetic target | CCAGTGGATCCAGCAACCTCCAACAACTGTCCTCTTTCTATCTTGAAATTAATATCTTTCAGG (SEQ ID NO:35) | 661000 |
| 1078delT | 2 | none | invader | AGTGCATAGGGAAGCACAGATAAAAACACCACAT (SEQ ID NO:36) | 413500 |
| 1078delT | 2 | hex | probe/DM | CGCGCCGAGGAGAACCCTGAGAAGAAGAA (SEQ ID NO:37) | 355400 |
| 1078delT | 2 | none | synthetic target | AGCCTTCTTCTTCTCAGGGTTCTTGTGGTGTTTTTATCTGTGCTTCCCTATGCACT (SEQ ID NO:38) | 533300 |
| G551D | 2 | none | invader | GCAGAGAAAGACAATATAGTTCTTGGAGAAGGTGGAATCACACTGAGTGGAGT (SEQ ID NO:39) | 628200 |
| G551D | 2 | hex | probe/DM | CGCGCCGAGGATCAACGAGCAAGAATTTCT (SEQ ID NO:40) | 343800 |
| G551D | 2 | none | synthetic target | CTTGCTAAAGAAATTCTTGCTCGTTGATCTCCACTCAGTGTGATTCCACCTTCTCCAAGAACTATATTGTCTTTCTCTGCAAACTT (SEQ ID NO:41) | 883100 |
| I148T | 1 | none | invader | AAATCAAACTAAACATAGCTATTCTCATCTGCATTCCAT (SEQ ID NO:42) | 432400 |
| I148T | 1 | hex | probe/ER24 | ACGGACGCGGAGGTGTGATGAAGGCCAAA (SEQ ID NO:43) | 350200 |
| I148T | 1 | none | synthetic target | CCATTTTTGGCCTTCATCACACTGGAATGCAGATGAGAATAGCTATGTTTAGTTTGATTT (SEQ ID NO:44) | 643100 |
| N1303K | 2 | none | invader | CCATATTTCTTGATCACTCCACTGTTCATAGGGATCCAAT (SEQ ID NO:45) | 414700 |
| N1303K | 2 | hex | probe/DM | CGCGCCGAGGCTTTTTTCTAAATGTTCCAGAAAAA (SEQ ID NO:46) | 391200 |
| N1303K | 2 | none | synthetic target | ATTTATTTTTTCTGGAACATTTAGAAAAAAGTTGGATCCCTATGAACAGTGGAGTGATCAAGAAATATGGAAAG (SEQ ID NO:47) | 867100 |
| 711+1G>T | 2 | none | invader | GCCTTTCCAGTTGTATAATTTATAACAATAGTGCCTAAAAGATTAAATCAATAGGTACATT (SEQ ID NO:48) | |
| 711+1G>T | 2 | hex | probe/DM | CGCGCCGAGGAATTCATCAAATTTGTTCAGGT (SEQ ID NO:49) | |
| 711+1G>T | 2 | none | synthetic target | ACCTGAACAAATTTGATGAATTATGTACCTATTGATTTAATCTTTTAGGCACTATTGTTATAAATTATACAACTGGAAAGGC (SEQ ID NO:50) | 927000 |
| 1717-1G>A | 3 | none | invader | GCCTTTCAAATTCAGATTGAGCATACTAAAAGTGACTCTCTAATTTTCTATTTTTGGTAATAT (SEQ ID NO:51) | 685000 |



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# Figure 2 cont'd

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|---|---|---|---|---|---|
| 1717-1G>A | 3 | hex | probe/DM | CGCGCCGAGGAGACATCTCCAAGTTTGC (SEQ ID NO:52) | 294500 |
| 1717-1G>A | 3 | none | synthetic target | CTCTGCAAACTTGGAGATGTCTTATTACCAAAAATAGAAAATTAGAGAGTCACTTTTAGTATGCTCAATCTGAATTTGAAAGGCACATC (SEQ ID NO:53) | 1010000 |
| W1282X | 3 | none | invader | GCTCACCTGTGGTATCACTCCAAAGGCTTTCCTA (SEQ ID NO:54) | 345000 |
| W1282X | 3 | hex | probe/DM | CGCGCCGAGGTCACTGTTGCAAAGTTATTG (SEQ ID NO:55) | 327800 |
| W1282X | 3 | none | synthetic target | GATTCAATAACTTTGCAACAGTGAAGGAAAGCCTTTGGAGTGATACCACAGGTGAGCAA (SEQ ID NO:56) | 683000 |
| 3849+10kbC>T | 2 | none | invader | CAAGAGTCTTCCATCTGTTGCAGTATTAAAATGGA (SEQ ID NO:57) | 390000 |
| 3849+10kbC>T | 2 | hex | probe/DM | CGCGCCGAGGTGAGTAAGACACCCTGAAA (SEQ ID NO:58) | 327400 |
| 3849+10kbC>T | 2 | none | synthetic target | TTCCTTTCAGGGTGTCTTACTCACCATTTTAATACTGCAACAGATGGAAGACTCTTG (SEQ ID NO:59) | 601000 |
| R553X | 4 | none | invader | CATTTACAGCAAATGCTTGCTAGACCAATAATTAGTTATTCACCTTGCTAAAGAAATTCTTGCTG (SEQ ID NO:60) | |
| R553X | 4 | hex | probe/DM | CGCGCCGAGGCATTGACCTCCACTCAGT (SEQ ID NO:61) | |
| R553X | 4 | none | synthetic target | ACTGAGTGGAGGTCAATGAGCAAGAATTTCTTTAGCAAGGTGAATAACTAATTATTGGTCTAGCAAGCATTTGCTGTAAATG (SEQ ID NO:62) | |
| G542X | 4 | none | invader | TCCAAGTTTGCAGAGAAAGACAATATAGTTCTTTC (SEQ ID NO:63) | |
| G542X | 4 | hex | probe/DM | CGCGCCGAGGGAGAAGGTGGAATCACA (SEQ ID NO:64) | |
| G542X | 4 | none | synthetic target | TGTGATTCCACCTTCTCAAAGAACTATATTGTCTTTCTCTGCAAACTTGGA (SEQ ID NO:65) | |
| 621+1G>T | 3 | none | invader | CCTTCATCACATTGGAATGCAGATGAGAATAGCTATGTTTAGTTTGATTTATAAGAAGC (SEQ ID NO:66) | 664000 |
| 621+1G>T | 3 | hex | probe/DM | CGCGCCGAGGTTAATACTTCCTTGCACAGG (SEQ ID NO:67) | 311900 |
| 621+1G>T | 3 | none | synthetic target | GGGGCCTGTGCAAGGAAGTATTAACTTCTTATAAATCAAACTAAACATAGCTATTCTCATCTGCATTCCAATGTGATGAAGGCCAA (SEQ ID NO:68) | 965000 |
| R334W | 2 | none | invader | CGCAGAACAATGCAGAATGAGATGGTGGTGAATATTTTCCT (SEQ ID NO:69) | 467000 |
| R334W | 2 | hex | probe/DM | CGCGCCGAGGAGAGGATGATTCCTTTGATTA (SEQ ID NO:70) | 336800 |
| R334W | 2 | none | synthetic target | TGCACTAATCAAAGGAATCATCCTCTGGAAAATATTCACCACCATCTCATTCTGCATTGTTCTGCG (SEQ ID NO:71) | 703000 |

| Mutation | Exemplary Pool | 3' blocking group | Oligo type (Arm) | Sequence (5'-3') | $\varepsilon_{260}$ $M^{-1}$ $cm^{-1}$ |
|---|---|---|---|---|---|
| Internal control | all | none | Invader | tgtacttcatgctgtctacactaagagagaatgagagacacaca (SEQ ID NO:72) | 503500 |
| Internal control | all | hex | Probe/SNP4b | tccgcgcgtcctgaagaagcaccaatcatg (SEQ ID NO:73) | 321200 |
| Internal control | all | none | Synthetic Target | tttcatgattggtgcttcttcagtgtgtctctcattctctcttagtgtagacagcatgaagtacattt (SEQ ID NO:74) | 698200 |

| | | | |
|---|---|---|---|
| all | Hex | DM/FAM | Y-tct-X-agc-cgg-ttt-tcc-ggc-tga-gac-ctc-ggc-gcg-hex (SEQ ID NO:75) |
| 1, 2, 5 | Hex | ER24/FAM | Y-tct-X-agc-cgg-ttt-tcc-ggc-tga-gac-tcc-gcg-tcc-gt-hex (SEQ ID NO:76) |
| all | Hex | SNP4b/Red | Y-tct-X-tcg-gcc-ttt-tgg-ccg-aga-gag-gac-gcg-cgg-a-hex (SEQ ID NO:77) |

X = Quencher = Z28
Y = Dye = FAM for 1055-48-08 and 1055-48-09 and Y = Z35 (or Redmond Red) for 1055-49-04

| Mutation | Pool | 3' blocking group | Oligo type (Arm) | Sequence (5'-3') | $\varepsilon_{260}$ $M^{-1}$ $cm^{-1}$ |
|---|---|---|---|---|---|
| delF508 | delF508 | none | Invader | TGATGACGCTTCTGTATCTATATTCATCATAGGAAACACA (SEQ ID NO:78) | 441500 |
| delF508 | delF508 | Hex | WT Probe | CGCGCCGAGGCAAAGATGATATTTTCTTTAATGGT (SEQ ID NO:79) | 382200 |
| delF508 | delF508 | Hex | Mut Probe | AGCTCGTCCGACACAATAATATTTTCTTTAATGGTGCCA (SEQ ID NO:80) | 418100 |
| delF508 | delF508 | Hex | DM/FAM | Y-tct-X-agc-cgg-ttt-tcc-ggc-tga-gac-ctc-ggc-gcg-hex (SEQ ID NO:81) | 347150 |
| delF508 | delF508 | Hex | Wingra/Red | Y-tct-X-tcg-gcc-ttt-tgg-ccg-aga-gat-gtc-gga-cga-gct-hex (SEQ ID NO:82) | 390400 |
| delF508 | delF508 | none | WT Target | TGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTTCCTATGATGAATATAGATACAGAAGCGTCATCAAA (SEQ ID NO:83) | 837500 |
| delF508 | delF508 | none | Mut Target | ATGCCTGGCACCATTAAAGAAAATATCATTGGTGTTTCCTATGATGAATATAGATACAGAAGCGTCATCAAA (SEQ ID NO:84) | 828100 |

| Mutation | Pool | 3' blocking group | Oligo type (Arm) | Sequence (5'-3') |
|---|---|---|---|---|
| 2184delA | 2184delA | none | Invader | CTTCCTTTTTTCCCCAAACTCTCCAGTCTGTTTAAAAGATTGTTTA (SEQ ID NO:85) |
| 2184delA | 2184delA | hex | MUT probe/DM | CGCGCCGAGGTTTGTTTCTGTCCAGGAG (SEQ ID NO:86) |
| 2184delA | 2184delA | hex | WT probe/ER24 | ACGGACGCGGAGTTTTGTTTCTGTCCAGGAG (SEQ ID NO:87) |

X = Quencher = Z28
Y = Dye = FAM for 1055-48-08 and Y = Z35 (or Redmond Red) for 1144-16-02

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|---|---|---|---|---|
| 711+1G>T | 2 | hex | probe/ER24 | ACGGACGCGGAGAATTCATCAAATTTGTTCAGG (SEQ ID NO:88) |
| 3849+10kb | 2 | hex | probe/ER24 | ACGGACGCGGAGTGAGTAAGACACCCTGAAA (SEQ ID NO:89) |

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